Hitesh H Shah

List of Publications by Year
in descending order

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		331670	434195
94	2,681	21	31
papers	citations	h-index	g-index

95	95	95	2608
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Does early and aggressive management of significant extrusion of the femoral head affect the outcome of Perthesâ€™ disease with the age of onset younger than 7Âyears?. Musculoskeletal Surgery, 2022, 106, 325-335.	1.5	3
2	Pseudoachondroplasia: Phenotype and genotype in 11 Indian patients. American Journal of Medical Genetics, Part A, 2022, 188, 751-759.	1.2	1
3	An unusual presentation, novel treatment with Meropenem PMMA beads and complications of Klebsiella osteomyelitis in a healthy adult- A case report. Journal of Clinical Orthopaedics and Trauma, 2022, 24, 101719.	1.5	1
4	Does the Deformity Index Reliably Predict the Shape of the Femoral Head at Healing of Legg-CalvÃ©-Perthes Disease?. Journal of Pediatric Orthopaedics, 2022, 42, e163-e167.	1.2	0
5	Radiologic Outcomes of Bilateral and Unilateral Perthes Disease: A Comparative Cohort Study. Journal of Pediatric Orthopaedics, 2022, 42, e168-e173.	1.2	1
6	A homozygous hypomorphicBNIP1variant causes an increase in autophagosomes and reduced autophagic flux and results in a spondyloâ€epiphyseal dysplasia. Human Mutation, 2022, 43, 625-642.	2.5	3
7	A systematic review of maternal diabetes and congenital skeletal malformation. Congenital Anomalies (discontinued), 2022, , .	0.6	0
8	The â€œDiscoid Epiphysisâ€â€”An Uncommon Presentation of Legg-CalvÃ©-Perthes Disease. Journal of Pediatric Orthopaedics, 2022, 42, e570-e576.	1.2	2
9	Steel syndrome: Report of three patients, including monozygotic twins and review of clinical and mutation profiles. European Journal of Medical Genetics, 2022, 65, 104521.	1.3	1
10	Demographics and Clinical Presentation of Early-Stage Legg-CalvÃ©-Perthes Disease: A Prospective, Multicenter, International Study. Journal of the American Academy of Orthopaedic Surgeons, The, 2021, 29, e85-e91.	2.5	4
11	Valgus-impacted fracture of neck of femur in a 12-year-old child. BMJ Case Reports, 2021, 14, e240707.	0.5	0
12	Slipped capital femoral epiphysis in a healed Perthes hip. BMJ Case Reports, 2021, 14, e243977.	0.5	0
13	The spectrum of tibial pseudarthrosis with constriction band syndrome in children. Journal of Pediatric Orthopaedics Part B, 2021, Publish Ahead of Print, .	0.6	1
14	Clinical and genetic spectrum of 104 Indian families with central nervous system white matter abnormalities. Clinical Genetics, 2021, 100, 542-550.	2.0	12
15	Controversies in the management of pediatric neck femur fractures- a systematic review. Journal of Orthopaedics, 2021, 27, 92-102.	1.3	8
16	Demographic and Practice Variability Amongst Indian Centres in a Multicentre Prospective Observational Study on Developmental Dysplasia of the Hip. Indian Journal of Orthopaedics, 2021, 55, 1559-1567.	1.1	2
17	Reliability of 3 Radiologic Classifications for the Severity of the Developmental Dysplasia of the Hip in Children Older Than 4 Years. Journal of Pediatric Orthopaedics, 2021, Publish Ahead of Print, 23-29.	1.2	1
18	Validation of Pediatric Self-Report Patient-Reported Outcomes Measurement Information System (PROMIS) Measures in Different Stages of Legg-CalvÃ©-Perthes Disease. Journal of Pediatric Orthopaedics, 2020, 40, 235-240.	1.2	9

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19	Ageâ€•and genderâ€•related reference ranges for thromboelastography from a healthy Indian population. International Journal of Laboratory Hematology, 2020, 42, 180-189.	1.3	11
20	Biallelic variants p.Arg1133Cys and p.Arg1379Cys in COL2A1: Further delineation of phenotypic spectrum of recessive Type 2 collagenopathies. American Journal of Medical Genetics, Part A, 2020, 182, 338-347.	1.2	6
21	Comparison of plaster-of-Paris casts and Woodcast splints for immobilization of the limb during serial manipulation and casting for idiopathic clubfoot in infants. Bone and Joint Journal, 2020, 102-B, 1399-1404.	4.4	1
22	Congenital pseudoarthrosis of the radius in Neurofibromatosis Type 1: An entity not to be missed!. Journal of Orthopaedics, 2020, 22, 427-430.	1.3	4
23	Surgical management of the congenital dislocation of the knee and hip in children presented after six months of age. International Orthopaedics, 2020, 44, 2635-2644.	1.9	6
24	Orthopedic manifestation as the presenting symptom of acute lymphoblastic leukemia. Journal of Orthopaedics, 2020, 22, 326-330.	1.3	6
25	Evolution of Legg-CalvÃ©-Perthes disease following proximal femoral varus osteotomy performed in the avascular necrosis stage: A prospective study. Journal of Children's Orthopaedics, 2020, 14, 58-67.	1.1	16
26	A rare case of rubber band syndrome of wrist with distal radius and ulna fracture. Journal of Orthopaedics, 2020, 20, 60-62.	1.3	1
27	Hemiepiphysiodesis using 2-holed reconstruction plate for correction of angular deformity of the knee in children. Journal of Orthopaedics, 2020, 20, 54-59.	1.3	8
28	Does the timing of treatment affect complications of pediatric femoral neck fractures?. Journal of Orthopaedics, 2020, 22, 207-212.	1.3	6
29	Management of Osteogenesis Imperfecta in India. , 2020, , 265-285.		0
30	An emerging ribosomopathy affecting the skeleton due to biallelic variations in NEPRO. American Journal of Medical Genetics, Part A, 2019, 179, 1709-1717.	1.2	3
31	Growth, sexual development and menstrual issues among girls with cerebral palsy â€“ A cross sectional study in a tertiary care centre. Clinical Epidemiology and Global Health, 2019, 7, 367-371.	1.9	2
32	Phenotyping and genotyping of skeletal dysplasias: Evolution of a center and a decade of experience in India. Bone, 2019, 120, 204-211.	2.9	10
33	Management of the Knee Problems in Spastic Cerebral Palsy. Indian Journal of Orthopaedics, 2019, 53, 53-62.	1.1	12
34	Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. Bone, 2018, 110, 368-377.	2.9	38
35	What Factors Influence Union and Refracture of Congenital Pseudarthrosis of the Tibia? A Multicenter Long-term Study. Journal of Pediatric Orthopaedics, 2018, 38, e332-e337.	1.2	24
36	Seven additional families with spondylocarpotarsal synostosis syndrome with novel biallelic deleterious variants in FLNB. Clinical Genetics, 2018, 94, 159-164.	2.0	10

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37	Pycnodysostosis: Novel Variants in CTSK and Occurrence of Giant Cell Tumor. Journal of Pediatric Genetics, 2018, 07, 009-013.	0.7	6
38	Knowledge among Mothersâ€™ of Children and Youth with Hemophilia-A Cross Sectional Survey at a Hemophilia Center. Indian Journal of Public Health Research and Development, 2018, 9, 27.	0.0	0
39	A review of skeletal dysplasia research in India. Journal of Postgraduate Medicine, 2018, 64, 98-103.	0.4	2
40	A novel sequence variant in SFRP4 causing Pyle disease. Journal of Human Genetics, 2017, 62, 575-576.	2.3	27
41	Second family provides further evidence for causation of Steel syndrome by biallelic mutations in <scp>COL27A1</scp>. Clinical Genetics, 2017, 92, 323-326.	2.0	18
42	A Rare Case of Congenital Tibiofemoral Fusion with Bilateral Proximal Femoral Focal Deficiency. JBJS Case Connector, 2017, 7, e22-e22.	0.3	0
43	Identification of a novel LRRK1 mutation in a family with osteosclerotic metaphyseal dysplasia. Journal of Human Genetics, 2017, 62, 437-441.	2.3	33
44	Fracture neck of femur in Factor XIII deficiency: Was better outcome possible?. Journal of Family Medicine and Primary Care, 2017, 6, 651.	0.9	0
45	Late presented case of distal humerus epiphyseal separation in a newborn. BMJ Case Reports, 2016, 2016, bcr2016215296.	0.5	7
46	Tibial hypoplasia with a bifid tibia: an unclassified tibial hemimelia. BMJ Case Reports, 2016, 2016, bcr2016216622.	0.5	2
47	Unclassified tibial hemimelia. BMJ Case Reports, 2016, 2016, bcr2016215305.	0.5	2
48	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. American Journal of Medical Genetics, Part A, 2016, 170, 410-417.	1.2	31
49	Phenotype and genotype in patients with Larsen syndrome: clinical homogeneity and allelic heterogeneity in seven patients. BMC Medical Genetics, 2016, 17, 27.	2.1	18
50	A novel multiple joint dislocation syndrome associated with a homozygous nonsense variant in the EXOC6B gene. European Journal of Human Genetics, 2016, 24, 1206-1210.	2.8	16
51	Post-operative Hypertension following Correction of Flexion Deformity of the Knees in a Spastic Diplegic Child: A Case Report. Malaysian Orthopaedic Journal, 2016, 10, 46-48.	0.5	0
52	Posttraumatic Static Volar Intercalated Segment Instability - Iatrogenic or Missed Injury. Journal of Orthopaedic Case Reports, 2016, 6, 59-61.	0.1	3
53	Novel and recurrent mutations in WISP3 and an atypical phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 2481-2484.	1.2	21
54	Mutations in patients with osteogenesis imperfecta from consanguineous Indian families. European Journal of Medical Genetics, 2015, 58, 21-27.	1.3	37

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55	Recurrent and novel GLB1 mutations in India. Gene, 2015, 567, 173-181.	2.2	22
56	Clinical utility of fetal autopsy and its impact on genetic counseling. Prenatal Diagnosis, 2015, 35, 685-691.	2.3	34
57	Apache Tez. , 2015, , .		138
58	A rare combination of amniotic constriction band with osteogenesis imperfecta. BMJ Case Reports, 2015, 2015, bcr2015212400-bcr2015212400.	0.5	3
59	Thorn-induced pseudotumour of the fibula. BMJ Case Reports, 2014, 2014, bcr2014204115-bcr2014204115.	0.5	2
60	A syndrome of facial dysmorphism, cubital pterygium, short distal phalanges, swan neck deformity of fingers, and scoliosis. American Journal of Medical Genetics, Part A, 2014, 164, 1035-1040.	1.2	5
61	The Sphericity Deviation Score. Journal of Pediatric Orthopaedics, 2014, 34, 522-528.	1.2	22
62	Perthes Disease. Orthopedic Clinics of North America, 2014, 45, 87-97.	1.2	37
63	A novel mutation (g.106737G>T) in zone of polarizing activity regulatory sequence (ZRS) causes variable limb phenotypes in Werner mesomelia. American Journal of Medical Genetics, Part A, 2014, 164, 898-906.	1.2	15
64	GALNS mutations in Indian patients with mucopolysaccharidosis IVA. American Journal of Medical Genetics, Part A, 2014, 164, 2793-2801.	1.2	31
65	Mutation spectrum of COL1A1 and COL1A2 genes in Indian patients with osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2014, 164, 1482-1489.	1.2	24
66	Exome Sequencing Identifies a Dominant <i>TNNT3</i> Mutation in a Large Family with Distal Arthrogryposis. Molecular Syndromology, 2014, 5, 218-228.	0.8	11
67	Does proximal femoral varus osteotomy in Leggâ€“CalvÃ©â€“Perthes disease predispose to angular mal-alignment of the knee? A clinical and radiographic study at skeletal maturity. Journal of Children's Orthopaedics, 2013, 7, 205-211.	1.1	15
68	Apache Hadoop YARN. , 2013, , .		1,314
69	The fate of the joint space in Leggâ€“CalvÃ©â€“Perthesâ€™ disease. Skeletal Radiology, 2013, 42, 341-345.	2.0	6
70	Quantitative Measures for Evaluating the Radiographic Outcome of Legg-CalvÃ©-Perthes Disease. Journal of Bone and Joint Surgery - Series A, 2013, 95, 354-361.	3.0	38
71	Talonavicular Arthrodesis for the Treatment of Neurological Flat Foot Deformity in Pediatric Patients. Journal of Pediatric Orthopaedics, 2013, 33, e39.	1.2	0
72	Further characterization of acro-renal-uterine-mandibular syndrome. Clinical Dysmorphology, 2012, 21, 83-86.	0.3	3

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73	A child with Erysipelothrix arthritisâ€“beware of the little known. Asian Pacific Journal of Tropical Biomedicine, 2012, 2, 503-504.	1.2	19
74	Duplications ofBHLHA9are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. Journal of Medical Genetics, 2012, 49, 119-125.	3.2	81
75	Growth retardation, intellectual disability, facial anomalies, cataract, thoracic hypoplasia, and skeletal abnormalities: A novel phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2941-2945.	1.2	2
76	Analysis of the WISP3 gene in Indian families with progressive pseudorheumatoid dysplasia. American Journal of Medical Genetics, Part A, 2012, 158A, 2820-2828.	1.2	63
77	Handedness in diplegic cerebral palsy. Developmental Neurorehabilitation, 2012, 15, 386-389.	1.1	3
78	Deletions in PITX1 cause a spectrum of lower-limb malformations including mirror-image polydactyly. European Journal of Human Genetics, 2012, 20, 705-708.	2.8	63
79	Loss of a condyle of the femur or tibia following septic arthritis in infancy: problems of management and testing of a hypothesis of pathogenesis. Journal of Children's Orthopaedics, 2012, 6, 319-325.	1.1	1
80	Quadricepsplasty for congenital dislocation of the knee and congenital quadriceps contracture. Journal of Children's Orthopaedics, 2012, 6, 397-410.	1.1	15
81	Environmental Tobacco and Wood Smoke Increase the Risk of Legg-CalvÃ©-Perthes Disease. Clinical Orthopaedics and Related Research, 2012, 470, 2369-2375.	1.5	23
82	The fate of the hip in spondylo-epi-metaphyseal dysplasia: clinical and radiological evaluation of adults with SEMD Handigodu type. Skeletal Radiology, 2012, 41, 939-945.	2.0	1
83	Congenital pseudarthrosis of the tibia: Management and complications. Indian Journal of Orthopaedics, 2012, 46, 616-626.	1.1	39
84	SP6-38 Tobacco smoke and the risk of Perthes' disease in south west India: a case-control study. Journal of Epidemiology and Community Health, 2011, 65, A465-A465.	3.7	0
85	Congenital Pseudarthrosis of the Tibia Treated With Intramedullary Rodding and Cortical Bone Grafting. Journal of Pediatric Orthopaedics, 2011, 31, 79-88.	1.2	35
86	Second report of slipped capital femoral epiphysis in Rubinsteinâ€“Taybi syndrome. Clinical Dysmorphology, 2011, 20, 55-57.	0.3	7
87	Hypoplasia/aplasia of pelvis, femora, fibulae, ulna, digits and nails. Clinical Dysmorphology, 2011, 20, 205-209.	0.3	0
88	Management of Severe Crouch Gait in Children and Adolescents With Cerebral Palsy. Journal of Pediatric Orthopaedics, 2010, 30, 832-839.	1.2	44
89	Primary protrusio acetabuli in childhood. Pediatric Radiology, 2010, 40, 55-55.	2.0	1
90	Donor site morbidity following the harvesting of cortical bone graft from the tibia in children. Journal of Children's Orthopaedics, 2010, 4, 417-421.	1.1	10

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91	Effect of Prophylactic Trochanteric Epiphyseodesis in Older Children With Perthes' Disease. Journal of Pediatric Orthopaedics, 2009, 29, 889-895.	1.2	35
92	Congenital posteromedial bowing of the tibia: a retrospective analysis of growth abnormalities in the leg. Journal of Pediatric Orthopaedics Part B, 2009, 18, 120-128.	0.6	33
93	To What Extent Does Remodeling of the Proximal Femur and the Acetabulum Occur Between Disease Healing and Skeletal Maturity in Perthes Disease?. Journal of Pediatric Orthopaedics, 2008, 28, 711-716.	1.2	19
94	Pre-axial mirror polydactyly associated with tibial deficiency: a study of the patterns of skeletal anomalies of the foot and leg. Journal of Children's Orthopaedics, 2007, 1, 49-54.	1.1	31